PARENT FACT SHEET

DISORDER

Homocystinuria (HCY)

CAUSE

HCY occurs when an enzyme called "cystathionine beta-synthase" (CBS) is either missing or not working properly. This enzyme's job is to break down methionine, an amino acid. When the CBS enzyme is not working correctly, methionine and another amino acid, homocystine, build up in the blood and cause problems.

IF NOT TREATED

Babies with HCY look healthy and normal at birth. If the condition is not treated, HCY can cause growth and learning delays, which are usually noticed between one and three years of age. HCY can also affect the eyes, bones, heart, and blood vessels.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is needed throughout life.

- The HCY infant will require a special formula, which will be prescribed by the metabolic specialist and dietician.
- As your child gets older, the dietician will develop a special food plan, which may include special medical foods.
- The metabolic specialist will prescribe medications for your child and the proper amounts to give. These will include Vitamin B6, Betaine, Vitamin B12, Folic Acid, and L-cystine.
- Do not make any changes to medications or diet without the approval and permission of the metabolic specialist and the dietician.
- Your child will require regular blood and urine tests to check their amino acid levels. Diet and medication changes may occur as a result.
- Contact your child's doctor immediately at the start of any illness. Children with HCY may need to be treated in a hospital to prevent serious health problems.

IF TREATED

With lifelong treatment, many children experience normal growth and learning abilities. Treatment may lower the risk of blood clots, heart disease and stroke. Treatment also decreases the chance of eye problems. Children who begin treatment later in life may have mental delays and behavior problems.